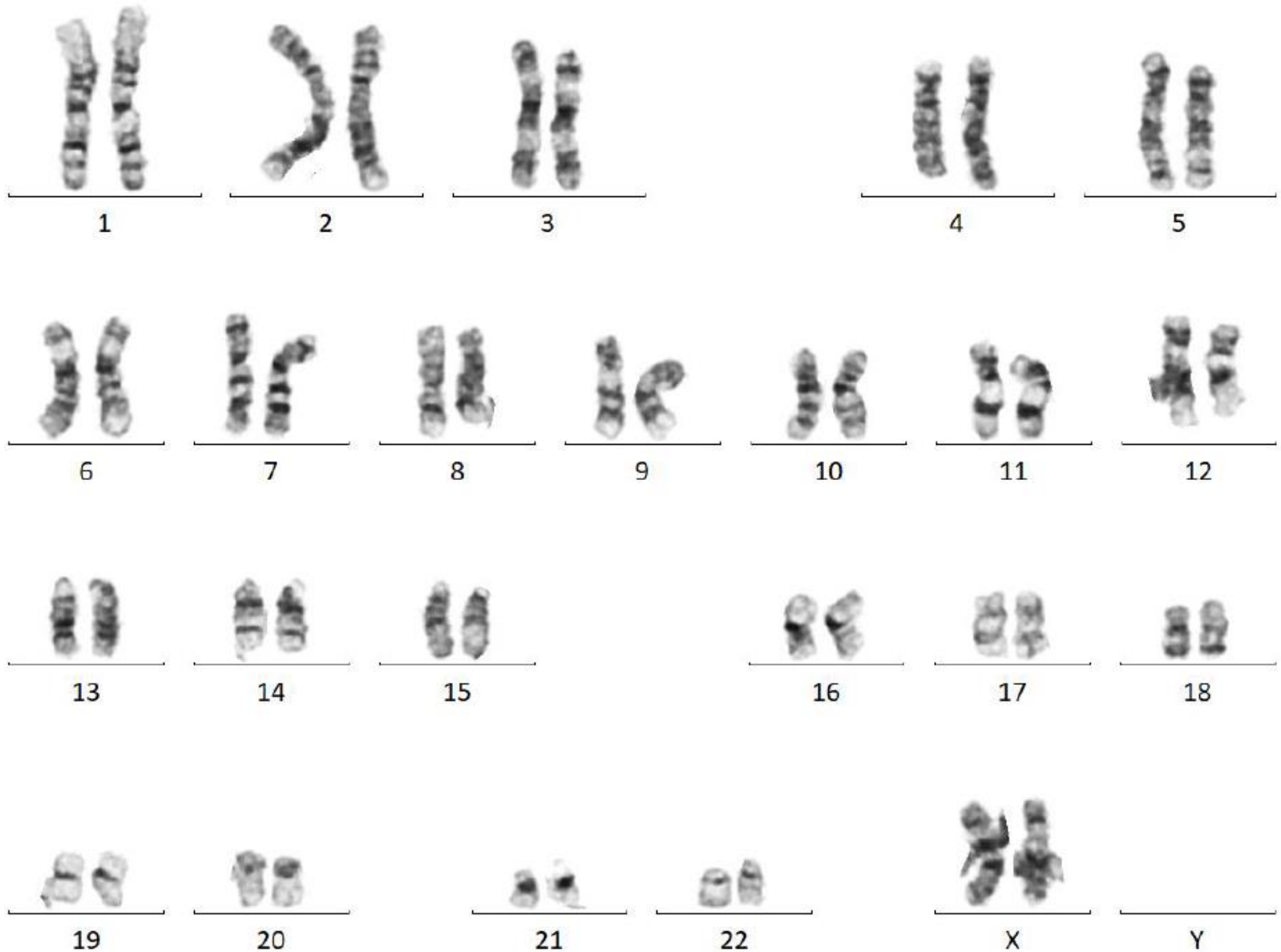
- You are to provide the ISCN nomenclature of the patient based on the karyotype given for selected metaphases. Consider that the remaining metaphases have a normal karyotype with the corresponding sex chromosomes. 20 metaphases have been analyzed in total.

- You are to determine if the variant identified is a reportable variant based on sequencing reads and quality metrics.
- You are to provide coding and protein nomenclature for the variant highlighted by the red arrow.
- You are to determine which variants are to be reported using the AMP/ASCO/CAP somatic variant classification scheme.
- Variant allele frequency of each candidate variant is to be noted on the report.
- Where applicable, suspicious germline variant(s) for germline sequencing should be identified.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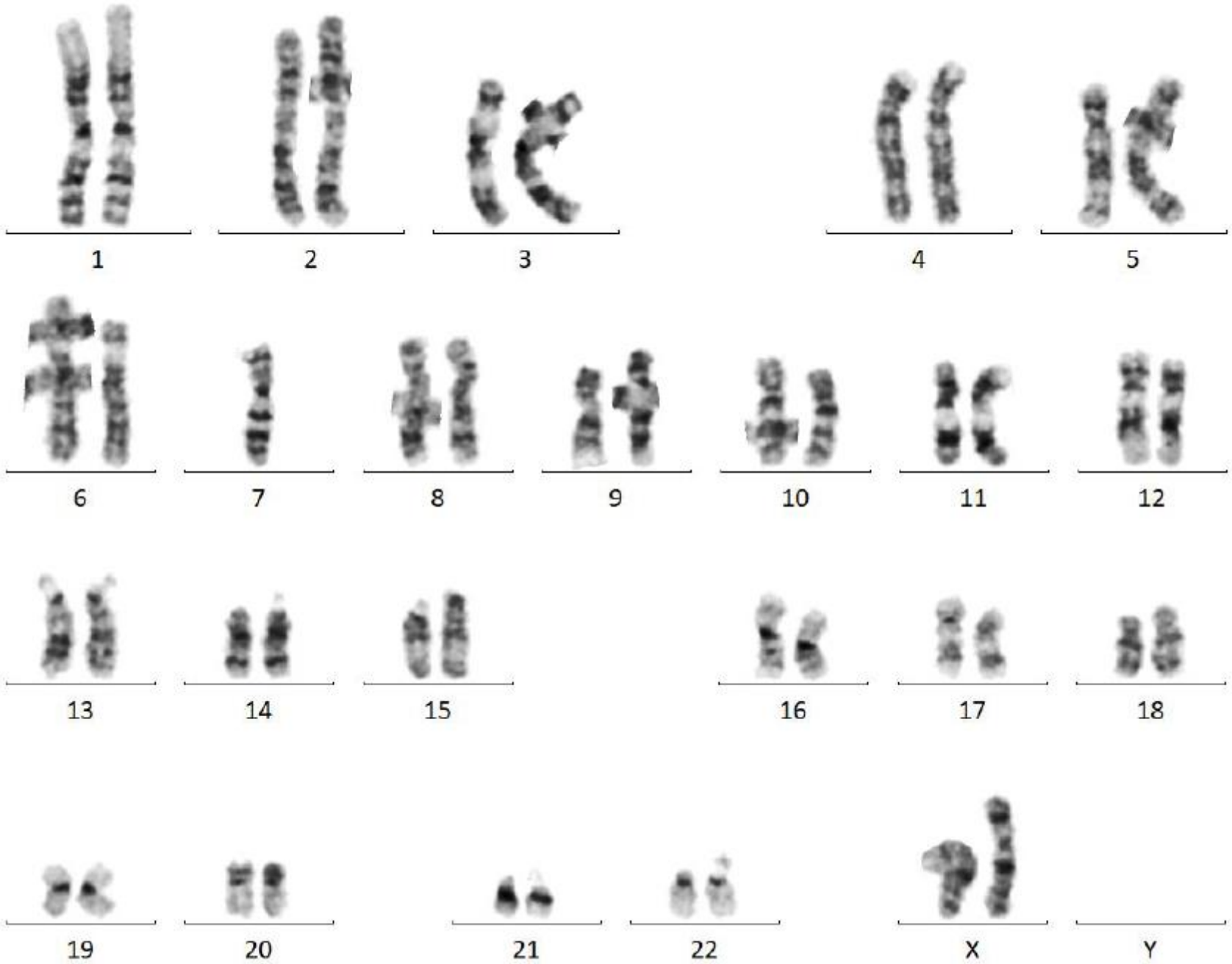
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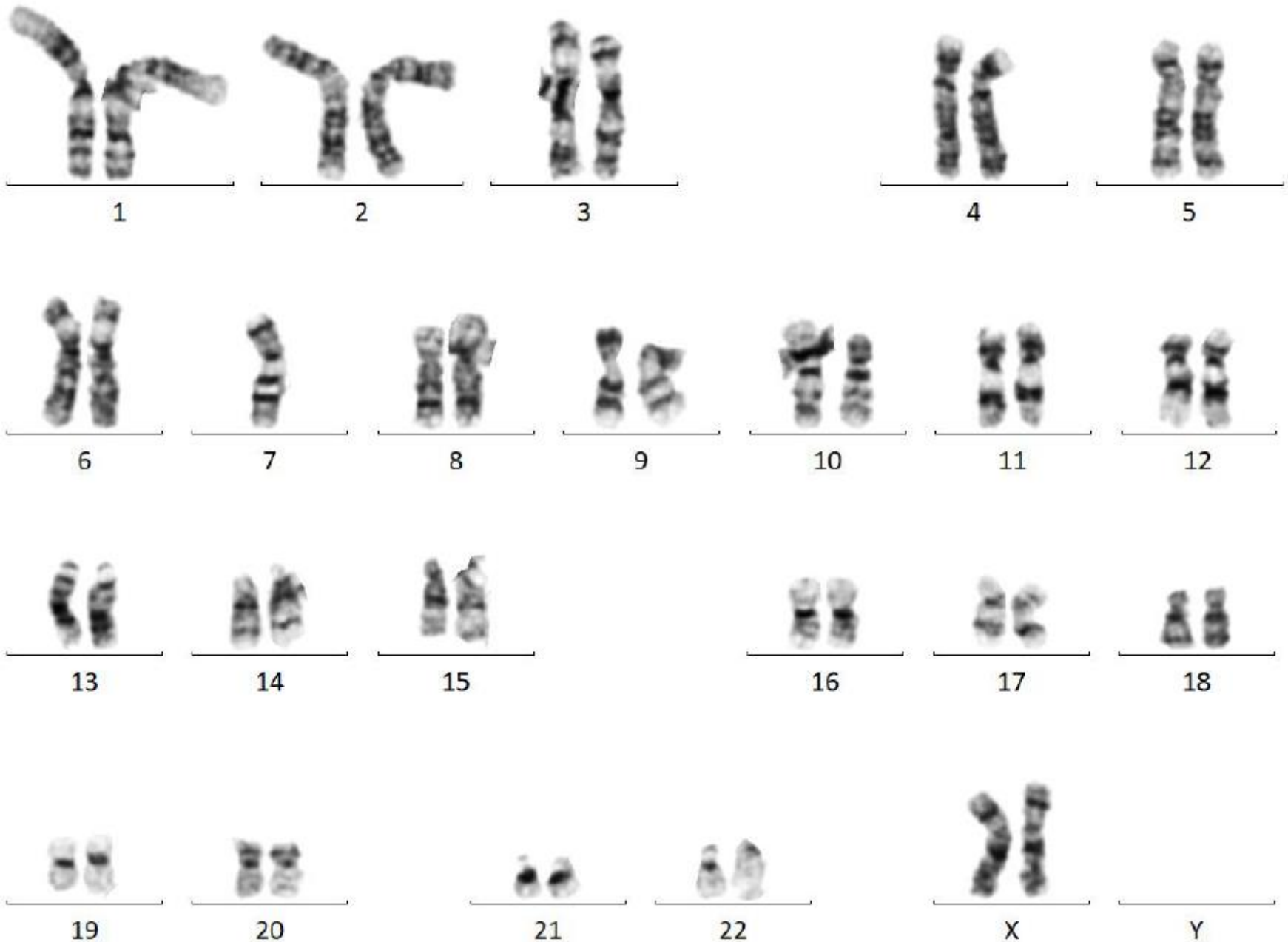
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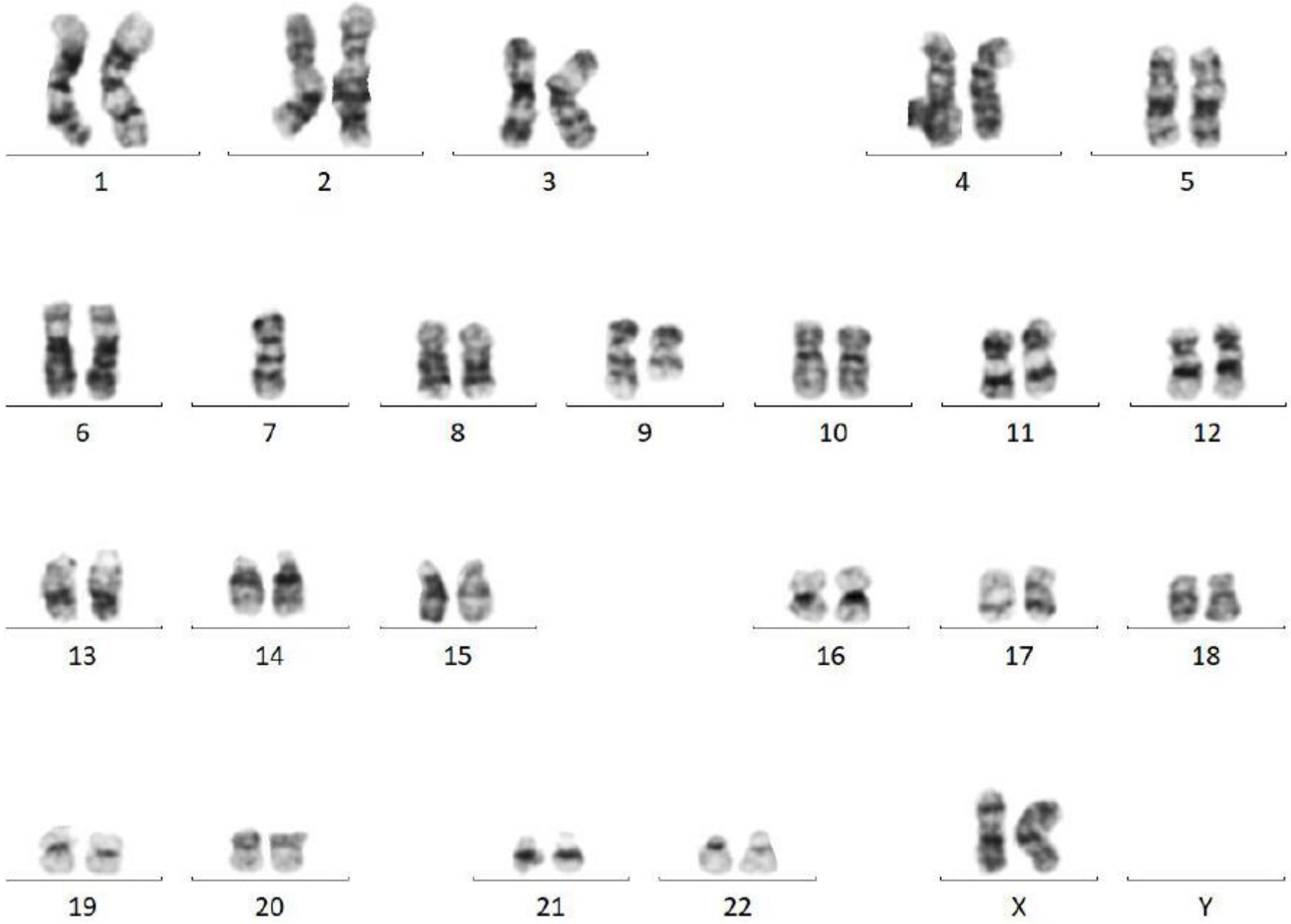
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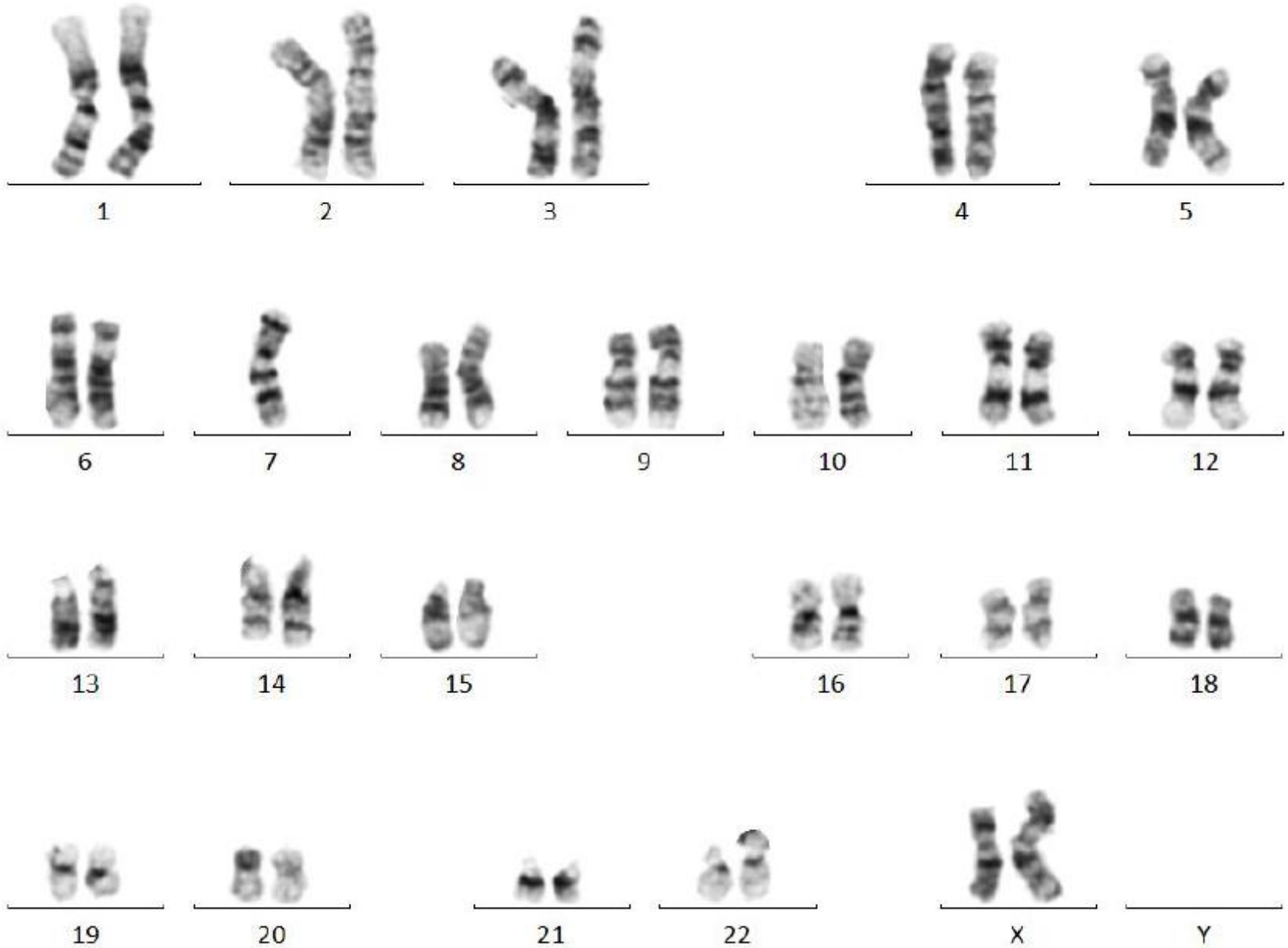
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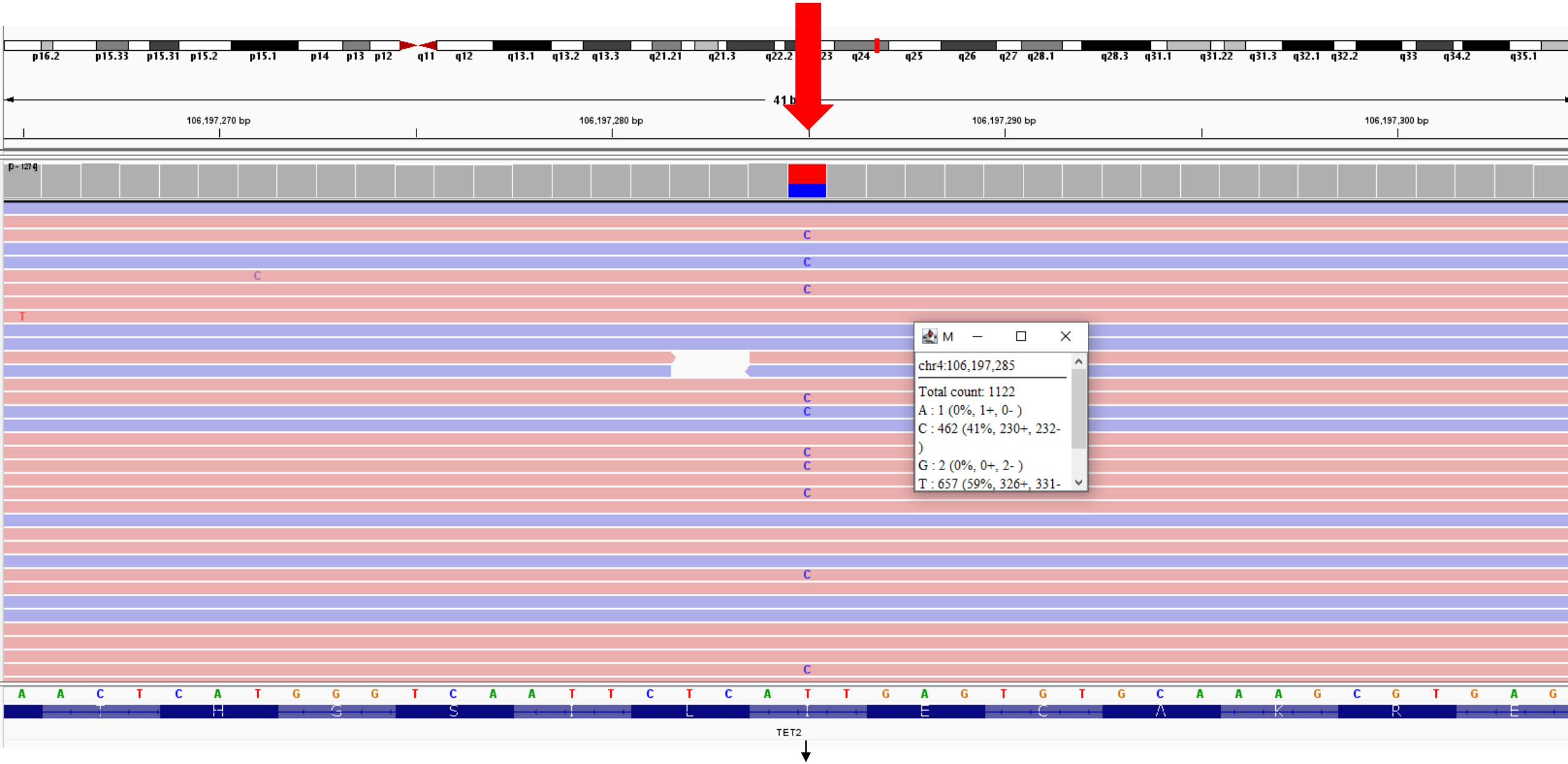
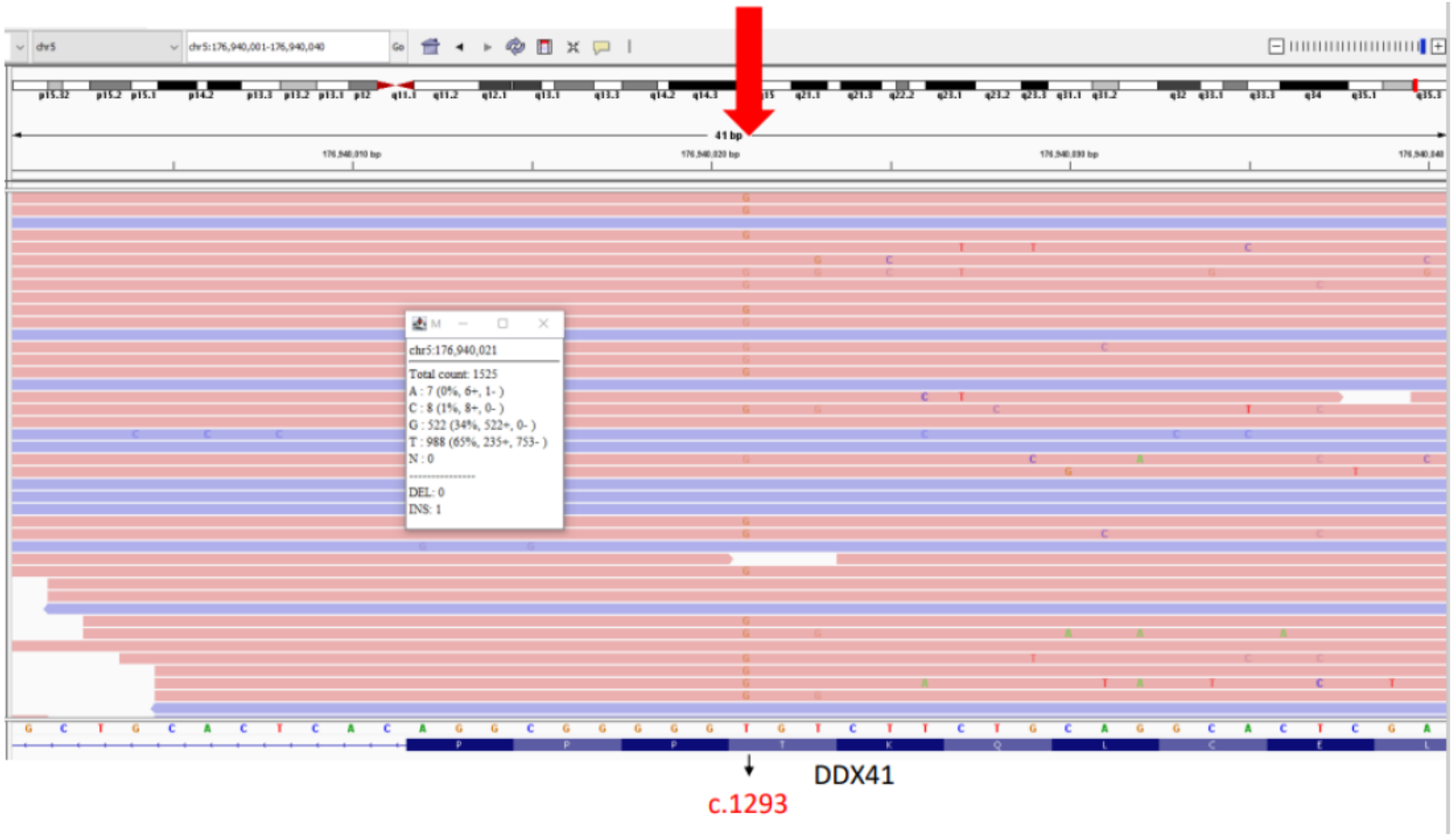
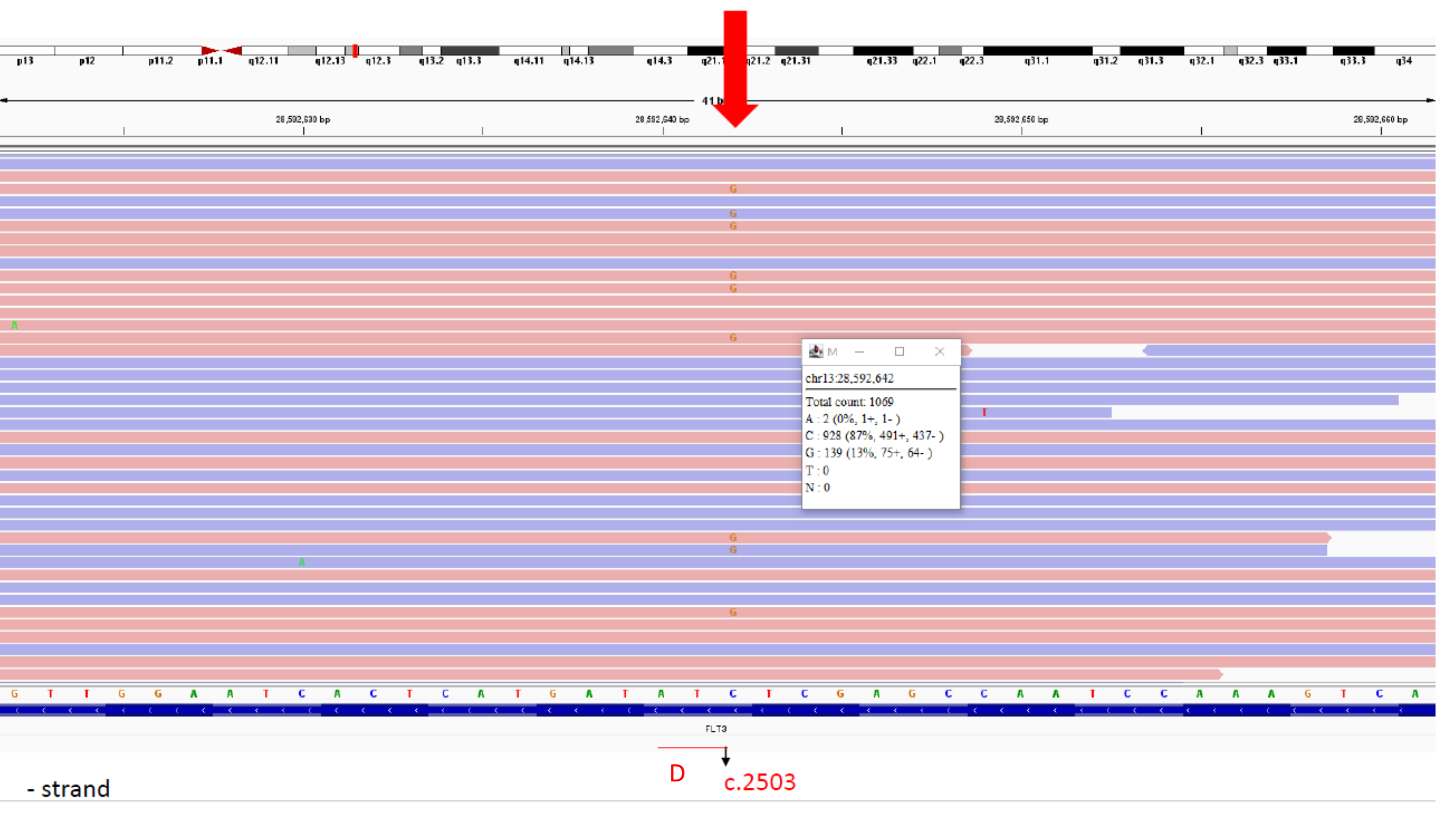


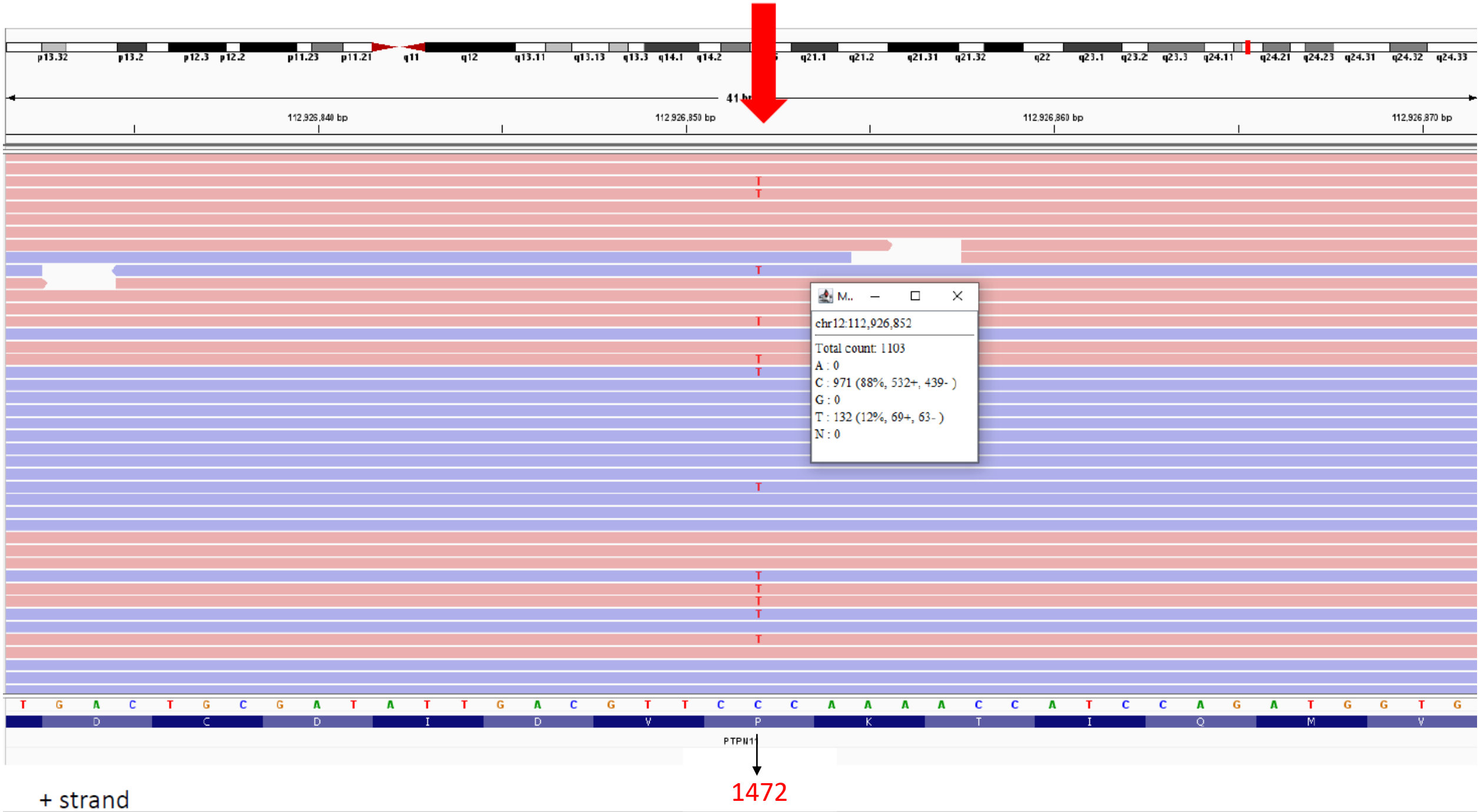
+ strand

c.5618


chr5
chr5:176,940,001-176,940,040
Go
41 bp
chr5:176,940,021
Total count: 1525
A : 7 (0%, 6+, 1-)
C : 8 (1%, 8+, 0-)
G : 522 (34%, 522+, 0-)
T : 988 (65%, 235+, 753-)
N : 0
DEL: 0
INS: 1
DDX41
c.1293

- strand


chr13:28,592,642
Total count: 1069
A : 2 (0%, 1+, 1-)
C : 928 (87%, 491+, 437-)
G : 139 (13%, 75+, 64-)
T : 0
N : 0
FLT3
D
c.2503
- strand


p13.32 p13.2 p12.3 p12.2 p11.23 p11.21 q11 q12 q13.11 q13.13 q13.3 q14.1 q14.2 q21.1 q21.2 q21.31 q21.32 q22 q23.1 q23.2 q23.3 q24.11 q24.21 q24.23 q24.31 q24.32 q24.33
41 bp
112,926,840 bp
112,926,850 bp
112,926,860 bp
112,926,870 bp
chr12:112,926,852
Total count: 1103
A : 0
C : 971 (88%, 532+, 439-)
G : 0
T : 132 (12%, 69+, 63-)
N : 0
T G A C T G C G A T A T T G A C G T T C C C A A A A C C A T C C A G A T G G T G
D C D I D V P K T I Q M V
PTPN11
1472
+ strand